BIOGRAPHICAL SKETCH

NAME	POSITION TITLE
Muenke, Maximilian	**Senior Investigator and Chief, Medical Genetics Branch**

EDUCATION / TRAINING

INSTITUTION AND LOCATION	DEGREE *(if applicable)*	MM/YY	FIELD OF STUDY
Goethe-Gymnasium Ibbenbueren, Germany	Abitur	1972	
Free University of Berlin School of Medicine Berlin, Germany	M.D.	1979	Medicine

Personal Statement:

I serve as the Chief of the Medical Genetics Branch of the Division of Intramural Research at the National Human Genome Research Institute, National Institutes of Health (NIH) and the Director of the NIH Medical Genetics and Genomic Medicine Residency and Fellowship programs. The focus of my laboratory's research has been on the delineation and identification of the underlying causes of craniofacial anomalies in humans: Opitz GBBB syndrome, craniosynostosis syndromes including the most common one, Muenke syndrome, congenital heart defects, and holoprosencephaly. More recently, our studies of the most common behavioral disorder of childhood, attention deficit hyperactivity disorder (ADHD) have led to the identification of ADHD and comorbid disorder susceptibility loci and genes.

Employment:

1978-79 Intern, Departments of Internal Medicine, Surgery and Pediatrics, Free University of Berlin
1979-80 Postdoctoral Associate, Department of Human Genetics, Free University of Berlin, Germany
1980-82 Pediatric Resident, Department of Pediatrics, Christian-Albrechts University, Kiel, Germany
1983-86 Postdoctoral Fellow, Department of Genetics, Yale University School of Medicine, New Haven, CT
1986-89 Postdoctoral Fellow in Clinical Genetics, The Children's Hospital of Philadelphia
1987-90 Associate, Howard Hughes Medical Institute and Department of Human Genetics, University of Pennsylvania School of Medicine, Philadelphia, PA
1990-96 Assistant Professor, University of Pennsylvania School of Medicine, Department of Pediatrics / The Children's Hospital of Philadelphia, Division of Human Genetics & Molecular Biology, Philadelphia, PA
1996-00 Associate Professor (with tenure), University of Pennsylvania School of Medicine, Departments of Pediatrics & Genetics, The Children's Hospital of Philadelphia, PA
2000- Senior Investigator and Chief, Medical Genetics Branch, NHGRI, NIH, Bethesda, MD

Other Experience and Professional Memberships:

1987 Medical Licensure, Pennsylvania
1990 Diplomate, American Board of Medical Genetics
1990 Certification in Clinical Genetics (American Board of Medical Genetics) (Lifetime certification)
1990 Certification in Clinical Cytogenetics (American Board of Medical Genetics) (Lifetime certification)
1993-2019 Certification in Clinical Molecular Genetics (American Board of Medical Genetics)
1994-97 Director, American Board of Medical Genetics-accredited Medical Genetics Fellowship Training Program of the Children's Hospital of Philadelphia and the University of Pennsylvania
1997- Director, NIH Medical Genetics and Genomic Medicine Residency and Fellowship Programs
2001-05 Member, Board of Directors, American Board of Medical Genetics
2007 Medical Licensure, Maryland
2010-13 Chair, John M. Opitz Young Investigator Award Committee
2010-14 Chair, Scientific Advisory Board, Freiburg Institute of Advanced Studies, Albert-Ludwig University Freiburg, Germany
2013- Founding Editor, *Molecular Genetics and Genomic Medicine*

Honors:

1983-85 Scholarship of the German Research Foundation
1991-93 Basil O'Connor Starter Scholar Research Award (March of Dimes)
1992-97 First Independent Research Support and Transition Award, NIH
1992-94 Ethel Brown Foerderer Fund for Excellence, University of Pennsylvania
1996 M.A. honoris causa, University of Pennsylvania
1996 FGFR3-associated craniosynostosis has been named: "Muenke Syndrome"
2005 Award of the National Attention Deficit Disorder Association (ADDA) for "Outstanding Work on the Genetics of Attention Deficit Hyperactivity Disorder"
2005 Merit Award of the NIH Office of the Director in "Recognition of Commitment and Contributions to the NIH Clinical Training Programs"
2006 Member, Association of American Physicians
2010 US Patent 8,003,406 B2: Methods for detecting Attention-Deficit/Hyperactivity Disorder

Selected publications (in chronological order, selected from 199 published or in press peer-reviewed publications, 78 book chapters / reviews, and 4 edited books / journal issues) (*h* Index: 57 in Scopus on November 25, 2014)

Gripp KW, Wotton D, Edwards MC, Roessler E, Ades L, Meinecke P, Richieri-Costa A, Zackai EH, Massagué J, Muenke M, Elledge S J: Mutations in TGIF, cause holoprosencephaly and link Nodal signaling to human neural axis determination. Nature Genet *25*:205-208, 2000.

Bamford R, Roessler E, Burdine RD, Saplakoglu U, dela Cruz J, Splitt M, Goodship JA, Towbin J, Bowers P, Ferrero GB, Marino B, Schier AF, Shen MM, Muenke M, Casey BM: Loss-of-function mutations in the *EGF-CFC* gene, *CFC1* are associated with human left-right laterality defects. Nature Genet *26*:365-369, & 501, 2000.

Goldmuntz E, Bamford R, Karkera JD, Dela Cruz J, Roessler E, Muenke M: *CFC1* mutations in patients with transposition of the great arteries and double outlet right ventricle. Am J Hum Genet *70*:776-780, 2002. PMCID: PMC384955

Roessler E, Du Y, Mullor JL, Casas E, Allen WP, Gillessen-Kaesbach G, Roeder ER, Ming JE, Ruiz i Altaba A, Muenke M: Loss-of-function mutations in the human *GLI2* gene cause pituitary anomalies and holoprosencephaly-like features. Proc Natl Acad Sci USA *100*:13424-13429, 2003. PMCID: PMC263830

Edison R, Berg K, Remaley A, Kelley R, Rotimi C, Stevenson RE, Muenke M: Adverse birth outcome among mothers with low serum cholesterol. Pediatrics *120*:723-733, 2007.

Karkera JD, Lee JS, Roessler E, Banerjee-Basu S, Ouspenskaia MV, Mez J, Goldmuntz E, Bowers P, Towbin J, Belmont J, Baxevanis AD, Schier AF, Muenke M: Loss-of-function mutations in the *Growth Differentiation Factor-1* (*GDF1*) are associated with congenital heart defects in humans. Am J Hum Genet *81*:987-994, 2007. PMCID: PMC2265655

Roessler E, Ouspenskaia MV, Karkera JD, Veléz JI, Kantipong A, Lacbawan F, Bowers P, Belmont JW, Towbin J, Goldmuntz E, Feldman B, Muenke M: Reduced NODAL signaling strength via mutation of several pathway members including FOXH1 is linked to human heart defects and holoprosencephaly. Am J Hum Genet *83*:18-29, 2008. PMCID: PMC2443854

Arcos-Burgos M, Jain M, Acosta MT, Shively S, Stanescu H, Wallis D, Domené S, Vélez JI, Karkera JD, Balog J, Berg K, Kleta R, Gahl WA, Roessler E, Long R, Lie J, Pineda D, Londoño AC, Palacio JD, Arbelaez A, Lopera F, Elia J, Hakonarson H, Johansson S, Knappskog PM, Haavik J, Ribases M, Cormand B, Bayes M, Casas M, Ramos T, Hervas A, Maher BS, Seitz C, Freitag CM, Palmason H, Meyer J, Romanos M, Renner T, Jacob C, Lesch K-P, Farone SV, Swanson J, Vortmeyer A, Bailey-Wilson J, Castellanos FX, Muenke M: A common variant of the lathrophilin 3 gene confers susceptibility to ADHD and predicts effectiveness of stimulant medication. Mol Psychiatr *15*:1053-1066, 2010. (Figures of article featured on journal cover).

Bae G, Domené S, Roessler E, Schachter K, Kang J-S, Muenke M, Krauss R: Holoprosencephaly-associated mutations in CDON result in defective interactions with other Hedgehog receptors. Am J Hum Genet 89:231-240, 2011.

Jain M, Vélez JI, Acosta MT, Balog J, Roessler E, Palacio LG, Pineda D, Londoño AC, Palacio JD, Arbelaez A, Lopera F, Elia J, Hakonarson H, Seitz C, Freitag CM, Palmason H, Meyer J, Romanos M, Walitza S, Hemminger U, Warnke A, Romanos J, Renner T, Jacob C, Lesch K-P, Swanson J, Castellanos FX, Bailey-Wilson J, Arcos-Burgos M, Muenke M: A cooperative interaction between LPHN3 and 11q doubles the risk for ADHD. Mol Psychiatr *17*:741-747, 2012.

Acosta MT, Vélez JI, Bustamante ML, Balog JZ, Arcos-Burgos M, Muenke M: A two-locus interaction between LPHN3 and 11q predicts ADHD severity and long-term outcome. Translational Psychiatr 1, e17; doi:10.1038/tp.2011.14; published online 5 July 2011. PMCID: PMC3309519

Solomon, B.D., Bear, K.A., Wyllie, A., Keaton, A.A., Dubourg, C., David, V., Mercier, S., Odent, S., Hehr, U., Paulussen, A., Clegg, N.J., Delgado, M.R., Bale, S.J., Lacbawan, F., Ardinger, H., Aylsworth, A., Bhengu, M.L., Braddock, S., Braddoch, S., Brookhyser, K., Burton, B., Gaspar, H., Grix, A., Horovitz, D., Kanetzke, D., Kayserili, H., Lev, D., Nikkel, S.M., Norton, M., Roberts, R., Saal, H., Schaefer, G.B., Schneider, A., Smith E.K., Sowry, E., Spence, M.A., Shalev, S., Steiner, C.E., Balog, J.Z., Hadley, D.W., Zhou, N., Pineda-Alvarez, D.E., Roessler, E., Muenke, M.: Genotypic and phenotypic analysis of 396 individuals with mutations in *Sonic Hedgehog*. J. Med. Genet. *49*:473-479, 2012. PMID: 22791840

Roessler, E., Hu, P., Hong, S.-K., Srivastava, K., Carrington, B., Sood, R., Petrykowska, H., Elnitski, L., Ribeiro, L.A., Richieri-Costa, A., Feldman, B., Odenwald, W.F., Muenke, M.: Unique alterations of an ultraconserved non-coding element in the 3'UTR of ZIC2 in holoprosencephaly. PLOS One *7*(7):e39026, 2012; published online 31 July 2012. PMCID: PMC3409191

Bear, K.A., Solomon, B.D., Antonini, S., Arnold, I.J.P., França, M.M., Gerkes, E.H., Grange, D.K., Hadley, D.K., Jääskeläinen, J., Paulo, S.S., Rump, P., Stratakis, C.A., Thompson, E.M., Willis, M., Winder, T.L., Jorge, A.A.L., Roessler, E., Muenke, M.: Pathogenic mutations in *GLI2* cause a specific phenotype that is distinct from holoprosencephaly. J. Med. Genet. April 17, 2014 (Epub ahead of print).

Editor of Books, Journal Issues:

Muenke M, Solomon BD, Odent S (eds): Holoprosencephaly. Am J Med Genet Part C: Semin. Med. Genet. *154C*, 2010.

Muenke M, Kress W, Collmann H, Solomon BD (eds.): Monographs in Human Genetics. *19*: Craniosynostoses: Molecular Genetics, Principles of Diagnosis and Treatment. Karger Publishing, Basel, Switzerland, vol.19:1-244, 2011.

Muenke M, Volkow N (eds.): Genetics of Substance Use Disorders and Addiction. *Human Genetics* vol. 131, 2012.

Muenke, M., Kruszka, P., Sable, C., Belmont, J. (eds.): Congenital Cardiovascular Anomalies: Molecular Genetics, Principles of Diagnosis and Treatment. Karger Publishing, Basel, Switzerland, 2015 (in press).

Research Support:

Research is supported through the Intramural Program of NHGRI, NIH (NIH employee).